FIGURE 1

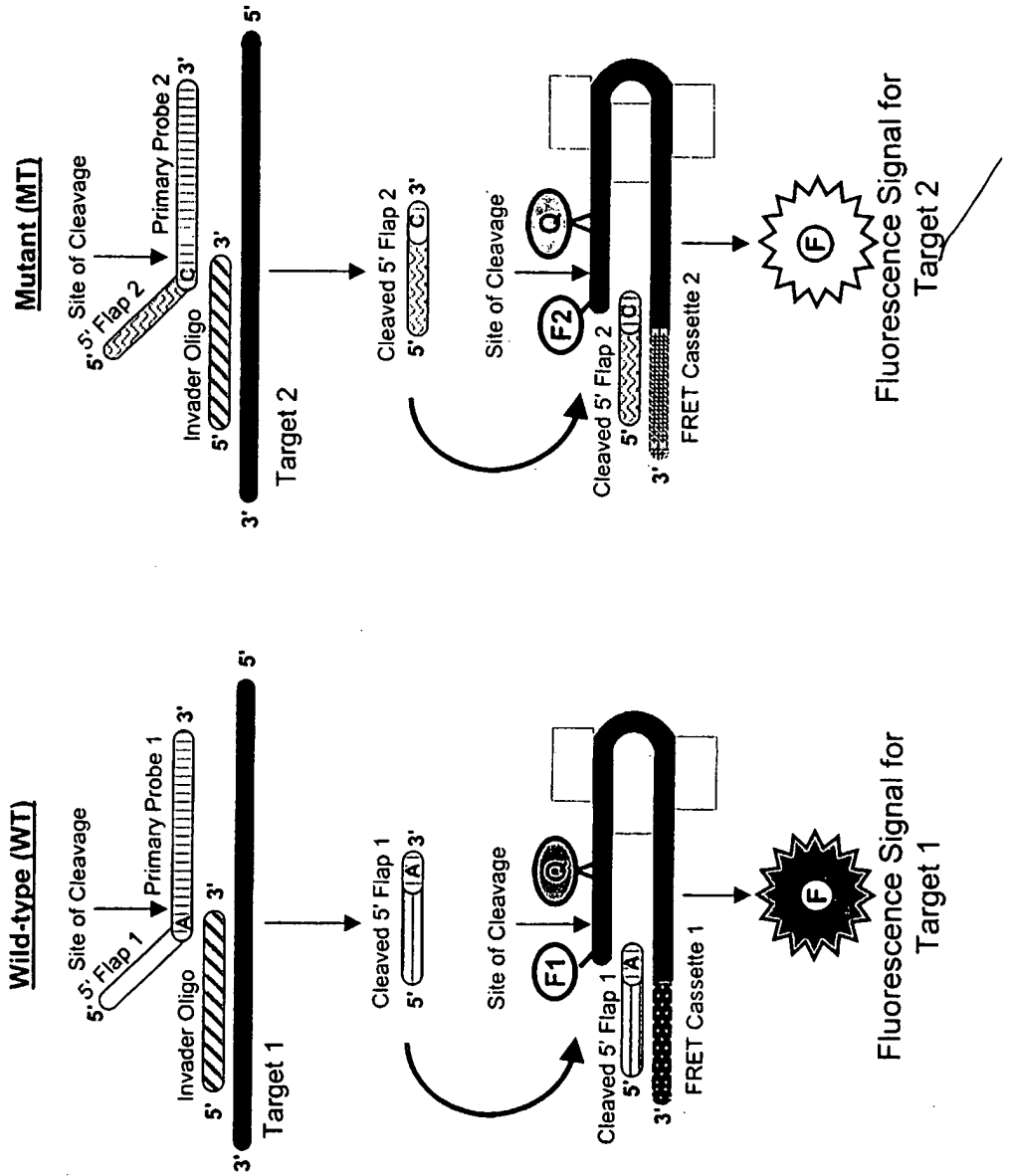

Wild-type (WT)
Site of Cleavage
5' Flap 1
Primary Probe 1
Invader Oligo
Target 1
Cleaved 5' Flap 1
Site of Cleavage
F1
Cleaved 5' Flap 1
FRET Cassette 1
F
Fluorescence Signal for Target 1
Mutant (MT)
Site of Cleavage
5' Flap 2
Primary Probe 2
Invader Oligo
Target 2
Cleaved 5' Flap 2
Site of Cleavage
F2
Cleaved 5' Flap 2
FRET Cassette 2
F
Fluorescence Signal for Target 2

FIGURE 2

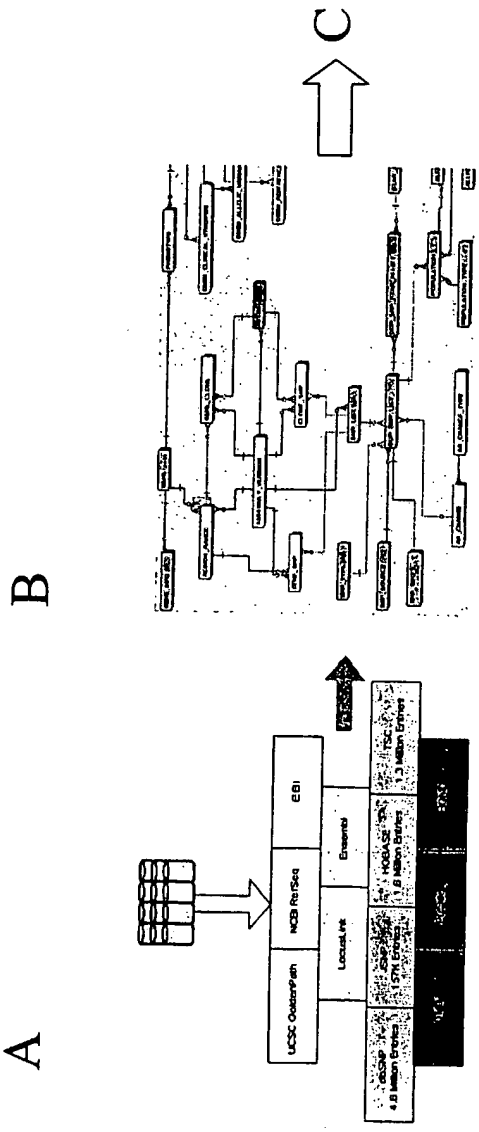


FIGURE 2 (continued)

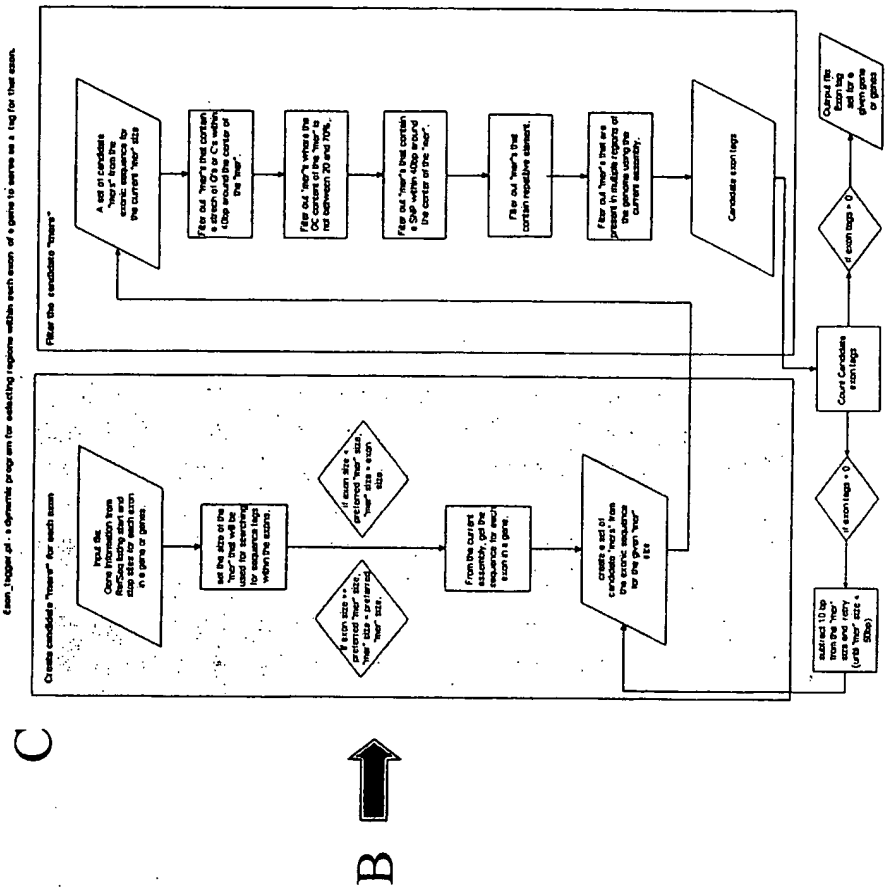


Figure 3

Chrom	Gene	%GC	Probe Oligo Sequence	SEQ ID NO	Invader Oligo Sequence	SEQ ID NO
1	ACTA1	N/A	ACGGACGCGGAGAGGAACCCTGTGACAT	1	CCATCCAGGGAAGAGTGGCCTGTTT	100
1	ACTA1	N/A	ACGGACGCGGAGAGGAACCCTGTGACATT	2	CCATCCAGGGAAGAGTGGCCTGTTT	101
1	ACTA1	N/A	ACGGACGCGGAGAGGAACCCTGTGACATTT	3	CCATCCAGGGAAGAGTGGCCTGTTT	102
1	ACTA1	N/A	ACGGACGCGGAGAGGAACCCTGTGACATTTC	4	CCATCCAGGGAAGAGTGGCCTGTTT	103
1	ACTA1	N/A	ACGGACGCGGAGGTGGCCTGTTAGGAAC	5	CGTTGGCACCCCATCCAGGGAAGAT	104
1	ACTA1	54%	ACGGACGCGGAGCCCCGCAGTCACT	6	GGGCGGCTGAGCTCCAGCCAT	105
1	ACTA1	58%	ACGGACGCGGAGTGGAGGTGGAGTGTG	7	TTCGTCGTCCTGAGAAGTCGCGTGCC	106
1	ACTA1	63%	ACGGACGCGGAGCGCGATCTCACCGA	8	GCGCCTGGACCTGGCGGGCT	107
1	ACTA1	62%	ACGGACGCGGAGGCCATTGTCGCACA	9	GCCGGCTTTCACCAGGCCGGAT	108
1	HIST2H2BE	58%	ACGGACGCGGAGGGTGACCGGACACA	10	GGCCCCATCGCACGGCAGAACTT	109
1	HIST2H2BE	56%	ACGGACGCGGAGCTGCGTGACAGCTC	11	ACAATTCAGCCCGGGTTCCGCAAACAA	110
1	SV2A	59%	ACGGACGCGGAGCAAGGGCAGCTGAG	12	CAGGGCCAGAGAGCTGCCAAGGGG	111
1	CRA	59%	ACGGACGCGGAGCTTCCCTGCTGGCA	13	TCAGCTCTGTCCTTGGCGGGACAGTCC	112
21	DSCR8		CGCGCCGAGGCAAGAAATTCTCATGTCTCAG	14	CACTGCAGCCTCCACCTCCCAGTTT	113
21	DSCR6	60%	CGCGCCGAGGCTCGACTCACGGCA	15	CCCAGGCTGGAATGCAATGGTGCGATT	114
21	DSCR8	52	CGCGCCGAGGCCGTGATTGAACCACTG	16	ACTGGGAGGTGGAGGCTGCAGTGATT	115
21	DSCR6	58	CGCGCCGAGGCTCCAGGTGTCTGGAT	17	GGCCTTCCAGTTCTGGTCAGCTCAGCATT	116
21	DSCR1	56	CGCGCCGAGGCAGTGAGCTCAGGAGA	18	GGTTCTGTCTGTGCCAAGGGCAGGTTGATT	117
21	AML1	58%	CGCGCCGAGGCCCACCTGTGCGA	20	GCCTCAGTGGAGACAAGTGGGAAAACATGGTT	119
21	AML1	56%	CGCGCCGAGGCCCTCTCTGCAGAACT	21	TGGACGTGCCAGCGGCATGACAAT	120
21	AML1	54%	CGCGCCGAGGCCTACCACAGAGCCA	22	CTTCACAAACCCACCGCAAGTCGCCAT	121
21	DSCR9	57%	CGCGCCGAGGGAGCAGTCTGTAACGTG	23	CTCCCCACCGTTCCATCCCAGGCT	122
21	DSCR9	57%	CGCGCCGAGGAGGCGAGCAGTCTG	24	GCACCTCCCCACCGTTCCATCCCT	123
X	L1CAM	58%	CGCGCCGAGGCCTGAGCAACGTGC	25	CGGATTCAGCGTGGCGCCCTGATT	124
X	L1CAM	58%	CGCGCCGAGGCCTGAGCAACGTGCA	26	CGGATTCAGCGTGGCGCCCTGATT	125
X	L1CAM	58%	CGCGCCGAGGCATGCAGTGCAGCAG	27	CGGATTCAGCGTGGCGCCCTGATCT	126
X	L1CAM	58%	CGCGCCGAGGGATCCTGAGCAACGTG	28	CCGGATTCAGCGTGGCGCCCTT	127
X	L1CAM	58%	CGCGCCGAGGGAGCAACGTGCAGC	29	GATTCAGCGTGGCGCCCTGATCCTT	128
X	L1CAM	58%	CGCGCCGAGGGTGCAGCCCAGTGA	30	CCGGATTCAGCGTGGCGCCCTT	129
X	PDCD8	52%	CGCGCCGAGGGCTACAAGCACGCTC	31	CAGATTTTGGTGGCTTCCGGGTAAATGCAGAT	130
X	PPEF1	53%	CGCGCCGAGGCGGTACCAGTTCTGGA	32	TGTCTGGCCTTCAGTCGAGCTTTGTAACCTT	131
Y	SRY	55%	CGCGCCGAGGCTCTGCAGCGAAGTG	33	GCTTCCCGCAGATCCCGCTTCGGTAT	132
Y	EIF1AY	56%	CGCGCCGAGGCTCTAAGAGCCGACTGA	34	GACCTCTTCCGACTCCTTTCTGGCGGTTACTAT	133
18	GATA6	54%	CGCGCCGAGGCGGCGTTTCTGCG	35	CACAAGCATTGCACACGGGTTCACCCTT	134
18	SERPINB2	57%	CGCGCCGAGGCCAGTTCTCCCTGTCA	36	CTGCCACAAACTGTGGGCCTCCATGTT	135
13	DLEU1	57%	CGCGCCGAGGTTCTGCGCATGTGC	37	AGGGAGAGCCGTGCACCACGATGAC	136
13	ABCC4	56%	CGCGCCGAGGAGGAGCACGTAGGTG	38	CGGCTGGCTGTGATCACACTGCCGT	137
13	POU4F1	65%	CGCGCCGAGGCCATCTGCAGGTTCG	39	CGTGGGCTCACTCAGCCAGAGCAT	138
13	POU4F1	65%	CGCGCCGAGGCTCCAGCTCCGAGG	40	CTGAGCACAAGTACCCGTCGCTGCAT	139
13	POU4F1	63%	CGCGCCGAGGGCCGAGAAACTGGAC	41	CCTCGTCCGAGAAGATCGCCGCCATCT	140
13	POU4F1	54%	CGCGCCGAGGGCCATAGGCCTCTCA	42	CCACTCACTTCCCGGGATTGGAGAGCAT	141

Aneuploidy Probe and Invader Designs

13	PCDH9	57%	CGCGCCGAGGCACTGTCACTGCAGC	43	ACTACCCACCGGCTCATGGTCTCCTAGACT	142
13	PCDH9	54%	CGCGCCGAGGCTGCACTCTGAGGCA	44	GGCCCTTTGTCTTGAAGCCTCCTTGGGAAT	143
13	PCDH9	54%	CGCGCCGAGGCCACTCCTGCTCGA	45	ACAGTGCTGGCTAGTGACGGCAGCTT	144
13	PCDH9	51%	CGCGCCGAGGTCCGTGGTAGCAGAA	46	GTGCCCCTCTCAGCCATTCCTGGCC	145
18	FLJ23403	52%	CGCGCCGAGGGCTGGATCTGTGTGG	47	TGGACGGACACAACTTTGAGCCTGTCCAT	146
18	KIAA0222	56%	CGCGCCGAGGGTGGAGACCAGAGTCA	48	CGAAGAGGTGACTTCGACCGAGCTCTCCAT	147
21	CLDN17	56%	CGCGCCGAGGCATGCCACAGATGCC	49	CGTTAGAGCCTGTGCACTGGACCTGCTTT	148
21	MGC33295	56%	CGCGCCGAGGCATGCTTTCGGAGACTC	50	CTCCCAGTCAGGCCTCCACCAGCT	149
21	PCP4	52%	CGCGCCGAGGCTGGTGCATCCATGTC	51	GGCCACCGCTGCACGTTCTGTCTT	150
X	PFKFB1	56%	CGCGCCGAGGCAGCTCCCTGAAGGT	52	TGGCCAACTTCATTCAGTCCCAGGGCATT	151
X	PFKFB1	56%	CGCGCCGAGGCAGCTCCCTGAAGGTG	53	TGGCCAACTTCATTCAGTCCCAGGGCATT	152
X	PFKFB1	56%	CGCGCCGAGGGATGCCCTGGGACT	54	GTGACTGGTCCACACCTTCAGGGAGCTT	153
X	ZNF157	60%	CGCGCCGAGGCCTTCCGTGCTGGA	55	GTTCACCCTGGGCCTCAGCCCAT	154
X	FLJ22843	62%	CGCGCCGAGGAGCTTCAACCGCAAC	56	CTGCCTCTGCTGCCGGGCAACT	155
X	DUSP21	59%	CGCGCCGAGGCATGCAGTGCAGCA	57	GAGGCGGAACGGCTCACTCCAGCT	156
X	DUSP21	59%	CGCGCCGAGGCATGCAGTGCAGCAG	58	GAGGCGGAACGGCTCACTCCAGCT	157
X	MGC33889	57%	CGCGCCGAGGCTGCGTAGCTCCCA	59	GGGTGACTCTCCTTCTGCCACAGTGGCT	158
Y	PRKY	48%	CGCGCCGAGGACGCCTGGATTATTCTG	60	TGTCTCCCCAGTCATTTCCCAAGAGAACACTCTCT	159
Y	PRKY	55%	CGCGCCGAGGGCCATGTGTTAGTGAAAATG	61	GCACCCAGAGTAGGGGTGGAGTATACCCTTT	160
Y	PRKY	53%	CGCGCCGAGGTAGCTCAGGTTCTCTGG	62	GTCCCCTCGGCTCCTAATCTCTTCTCAAAACTCAGC	161
Y	TMSB4Y	59%	CGCGCCGAGGGCAAGAAGGGCTGTG	63	TCGGCATCCCTAGCACACATGCCTCTT	162
Y	TMSB4Y	59%	CGCGCCGAGGCAGGTGGCTGTTTCTC	64	GCACAGTTCCCAGTACAGAGACCCGGAT	163
21	NRIP1	54%	CGCGCCGAGGCTGCCTTACCAGTGTC	65	GCTCCTGCTGTGCAGAGAAGCGCACT	164
21	NRIP1	54%	CGCGCCGAGGCTGCCTTACCAGTGTCC	66	GCTCCTGCTGTGCAGAGAAGCGCACT	165
21	HLCS	60%	CGCGCCGAGGGACATGCTGAGAAACCT	67	TGCACCCGGACGGCAACTCCTTCT	166
21	DSCR6	57%	CGCGCCGAGGCTGCTTCCGAAGCTG	68	GCCACGATTGACTTCTACGACGATGAGTCTACTGAGTT	167
21	DSCR9	58%	CGCGCCGAGGCTGTAACGTGGTGCAG	69	CCGTTCCATCCCAGGCGAGCAGTT	168
21	DSCR3	55%	CGCGCCGAGGGTCAACTGGCCACAG	70	AAGTGCGCTTGCAGCCATCCTGGATT	169
21	DSCR4	56%	CGCGCCGAGGCTTGCGGTCATGCAA	71	GTGGAAGGGTTTGTCTTGTGCCCAGGCT	170
21	DSCR4	56%	CGCGCCGAGGCTTGCGGTCATGCAAG	72	GTGGAAGGGTTTGTCTTGTGCCCAGGCT	171
21	DSCR10	59%	CGCGCCGAGGGCCATTTCCAGGTCC	73	TGGCCGAGCTCCACCGTGTCATCAT	172
18	CN2	54%	CGCGCCGAGGGTTGCTGCTGCAGAT	74	GGAGAAGAGAGGCGAAATCAGGAGGATGATGGAAT	173
21	HLCS	64%	CGCGCCGAGGCTTGAGCAGTGGCTG	75	ACCAGAGCGAGGTGAAGCTCAGCGTT	174
21	HLCS	64%	CGCGCCGAGGGACGCTGAGCTTCAC	76	TCCTGGTACCTGCAGCCACTGCTCAAT	175
21	HLCS	58%	CGCGCCGAGGCATGGCTCATGGACC	77	CGCTGCTTGAAGGCGTTTGCATGGTCT	176
21	DSCR9	57%	CGCGCCGAGGCGTCTGCGCACCT	78	TCCATTTTGCCAGGCGCTTGTCCATCT	177
21	DSCR9	57%	CGCGCCGAGGCGTCTGCGCACCTC	79	TCCATTTTGCCAGGCGCTTGTCCATCT	178
21	DSCR3	57%	CGCGCCGAGGTGAAGCTCTGCAGGA	80	ACCACCTCATCACGGAGAACTTCCCGCC	179
21	DSCR6	63%	CGCGCCGAGGGTAGTGTCCTGGCTCG	81	GCCTCTGCACACTCAGCAGGGATGCT	180
X	MTMR8	59%	CGCGCCGAGGCGGAAGATCTCAGTGCT	82	CCATGACTGCAGTTCCCGCCACT	181
X	MGC23947	53%	CGCGCCGAGGGATCCTGGGACCTCC	83	CTCCTTTTCTTCCCCCAGATGTCTGTGAACACAT	182
X	FLJ21174	56%	CGCGCCGAGGCAGCATCATGAGAGTCC	84	TGGCCTCCAGCTGGCATCTTGACCTT	183
X	ESX1L	62%	CGCGCCGAGGAGCTGCAGGAGCTAG	85	CCGCACCGCGTTCACGCAGTTTCT	184
X	ZNF157	52%	CGCGCCGAGGCAATGGTGAGGGAAGTC	86	CTGGTGATCATATGCCCCCAAGGAGCTTGATCT	185

Aneuploidy Probe and Invader Designs

X	PCTK1	57%	CGCGCCGAGGTGAAGCCTGGTCACT	87	CTGCACCTCATCCGAGGACGTGGCC	186
X	MGC33889	62%	CGCGCCGAGGTGCGTAGCTCCCAG	88	GGTGACTCTCCTTCTGCCACAGTGGCCC	187
21	DSCR9	57%	CGCGCCGAGGGAGCAGTCTGTAACGTG	89	CTCCCCACCGTTCCATCCCAGGCT	188
21	DSCR9	57%	CGCGCCGAGGAGGCGAGCAGTCTG	90	GCACCTCCCCACCGTTCCATCCCT	189
21	DSCR9	57%	CGCGCCGAGGCCATCCCAGGCGA	91	GTCTGCGCACCTCCCCACCGTTT	190
21	DSCR9	57%	CGCGCCGAGGTTCCATCCCAGGCG	92	CGTCTGCGCACCTCCCCACCGC	191
Y	SRY	55%	CGCGCCGAGGGACTGTACGAAAGCCAC	93	GAAGTGCAACTGGACAACAGGTTGTACAGGGATT	192
Y	SRY	55%	CGCGCCGAGGAGGTTGTACAGGGATGA	94	GGTACTCTGCAGCGAAGTGCAACTGGACAACT	193
Y	SRY	56%	CGCGCCGAGGGCAGATCCCGCTTC	95	TGCTGCCGAAGAATTGCAGTTTGCTTCCCT	194
Y	SRY	54%	CGCGCCGAGGGCTTCGGTACTCTGC	96	GAAGAATTGCAGTTTGCTTCCCGCAGATCCCT	195
21	CLDN17	59%	CGCGCCGAGGTGAAGCAGGTCCAGT	97	CGCCCTGCTTATTGGCATCTGTGGCAC	196
21	DSCR10	56%	CGCGCCGAGGGCATGGCTCTGCAC	98	GCTTGAAGGTGACACACAGTGACACGGGAT	197
21	MGC33295	57%	CGCGCCGAGGCCAGTCAGGCCTCC	99	GCAAAGTAAGGAGTCGCATCCCCTGCCTCT	198
21	STCH	54	CGCGCCGAGGCCACGTCTTGGTGATAG	217	GCCTATGGTCTCCACAAGGCTGACGTCTTT	218
21	STCH	53%	CGCGCCGAGGGCTTGGATAGCCACTC	219	GAGAGGCCAAGAGCCTCCATCAATCCCTT	220
X	AR	56%	CGCGCCGAGGCCAGTGCTCCGGA	221	CGACTCTGGTACGCAGCTGCCTCGTT	222
13	CCNA1	55%	CGCGCCGAGGGCGCATGCCTTCC	223	TCCACCAGCCAGTCCACCAGAATCGTT	224
13	CCNA1	49%	CGCGCCGAGGCGTAGGAACAGCAGC	225	GTCTGTTCTGAGAGGGAAACTGCAGCTT	226
13	ING1	52%	CGCGCCGAGGTCAGCGACCAATCGT	227	GCTAGGCCCGCCTTCATCCGCCC	228
21	ADAMTS5	58%	CGCGCCGAGGCCAGCAGTGCAACC	229	GAACTCCCAGGACAGACCTACGATGCCACT	230

FAM FRET cassette	Fam-TCT-Z28-AGCCGGTTTTCCGGCTGAGACCTCGGCGCG-hex	SEQ ID NO:199
RED FRET cassette	Red-TCT-Z28-TCGGCCTTTTGGCCGAGAGACTCCGCGTCCGT-hex	SEQ ID NO:200

FIGURE 4

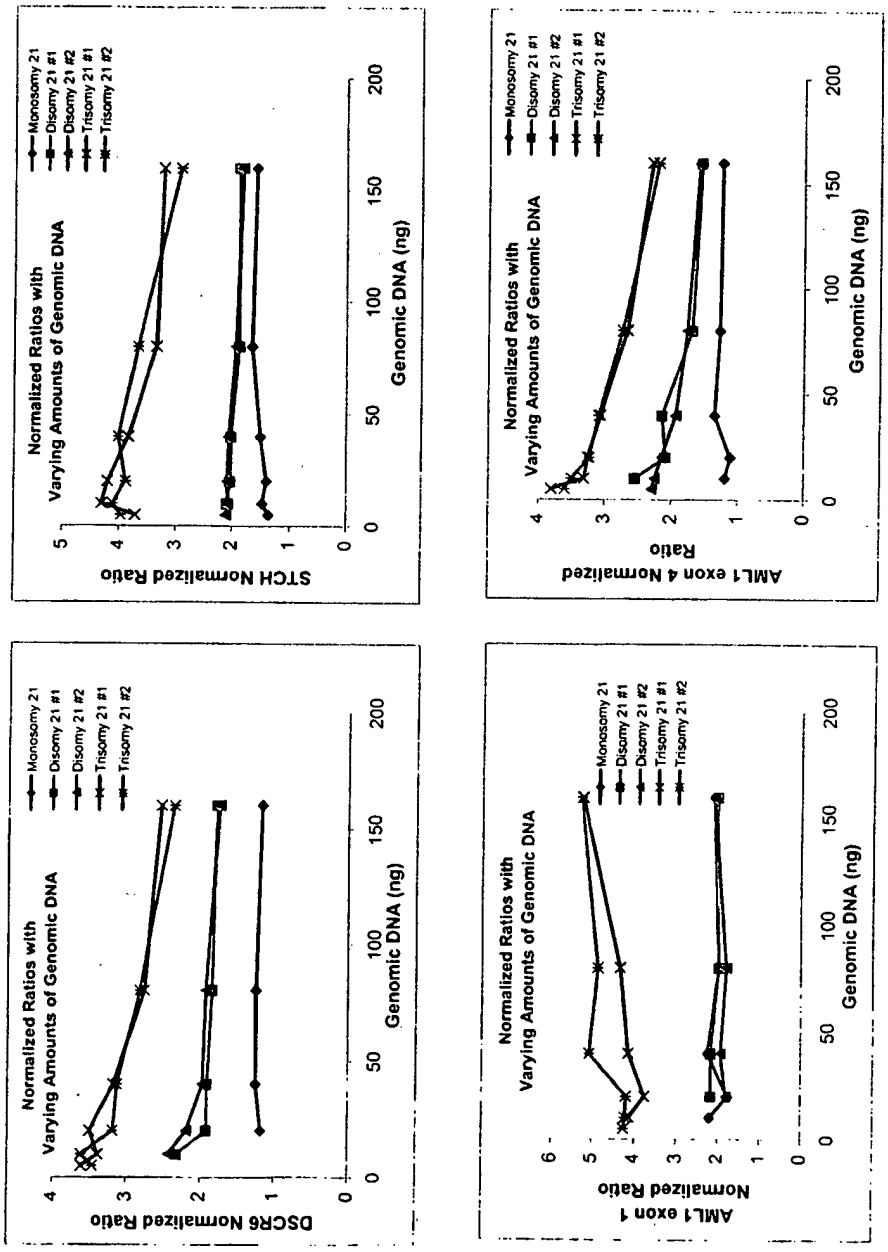

Normalized Ratios with Varying Amounts of Genomic DNA
Monosomy 21
Disomy 21 #1
Disomy 21 #2
Trisomy 21 #1
Trisomy 21 #2
DSCR6 Normalized Ratio
STCH Normalized Ratio
AML1 exon 1 Normalized Ratio
AML1 exon 4 Normalized Ratio
Genomic DNA (ng)
0
50
100
150
200
1
2
3
4
5
6

FIGURE 5

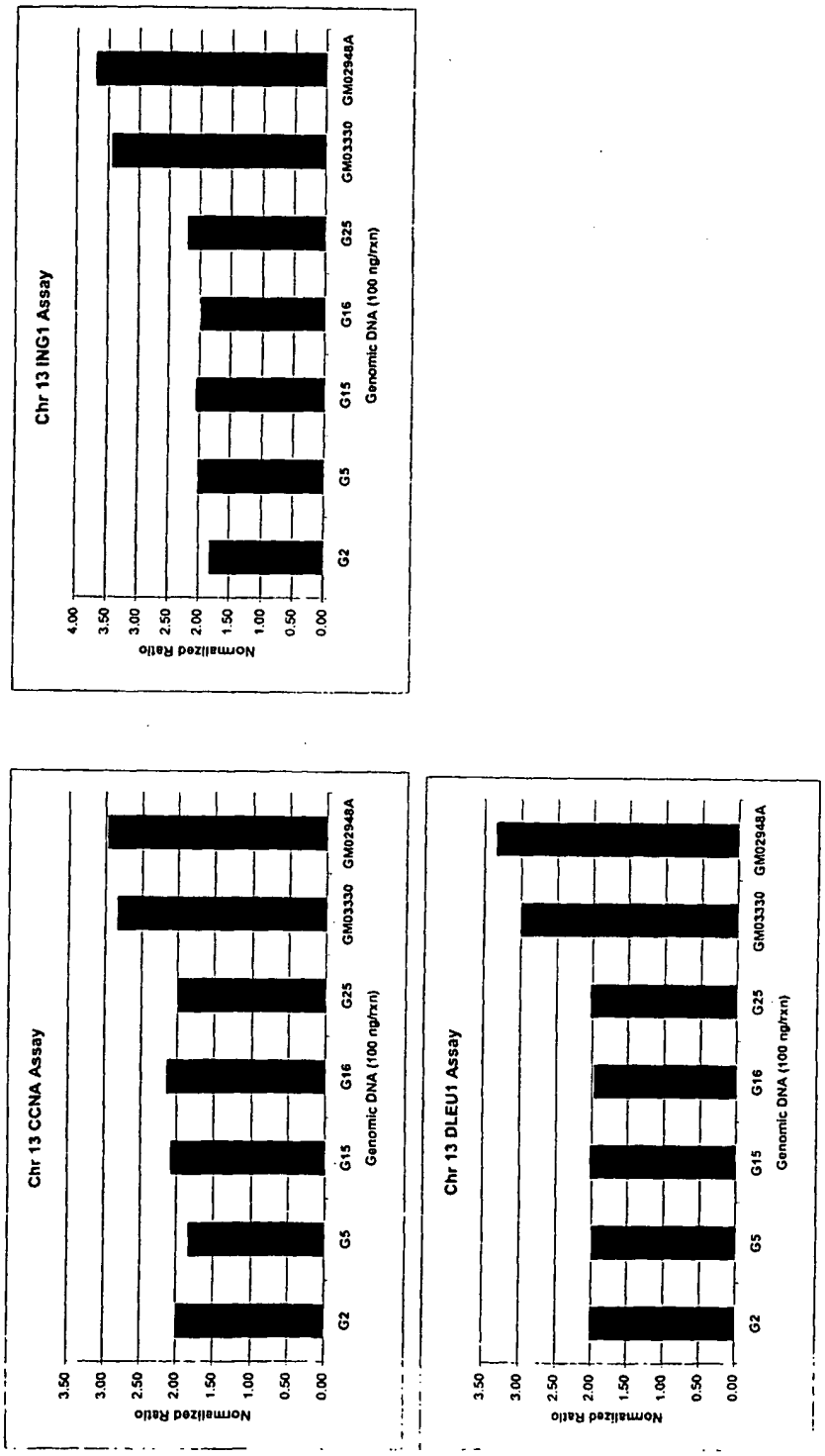
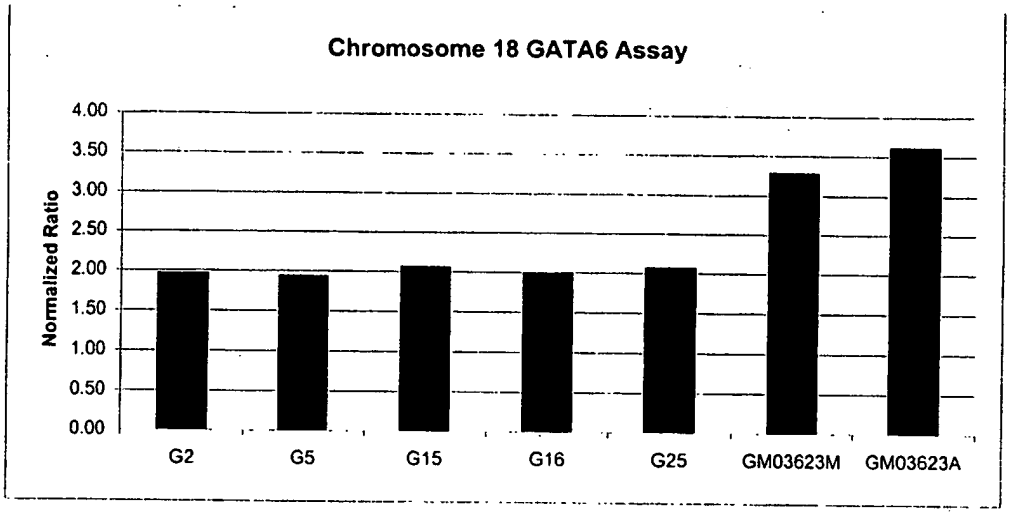

Chr 13 ING1 Assay
Normalized Ratio
4.00
3.50
3.00
2.50
2.00
1.50
1.00
0.50
0.00
G2
G5
G15
G16
G25
GM03330
GM02948A
Genomic DNA (100 ng/rxn)
Chr 13 CCNA Assay
Normalized Ratio
3.50
3.00
2.50
2.00
1.50
1.00
0.50
0.00
G2
G5
G15
G16
G25
GM03330
GM02948A
Genomic DNA (100 ng/rxn)
Chr 13 DLEU1 Assay
Normalized Ratio
3.50
3.00
2.50
2.00
1.50
1.00
0.50
0.00
G2
G5
G15
G16
G25
GM03330
GM02948A
Genomic DNA (100 ng/rxn)

FIGURE 6


Chromosome 18 GATA6 Assay
Normalized Ratio
4.00
3.50
3.00
2.50
2.00
1.50
1.00
0.50
0.00
G2
G5
G15
G16
G25
GM03623M
GM03623A

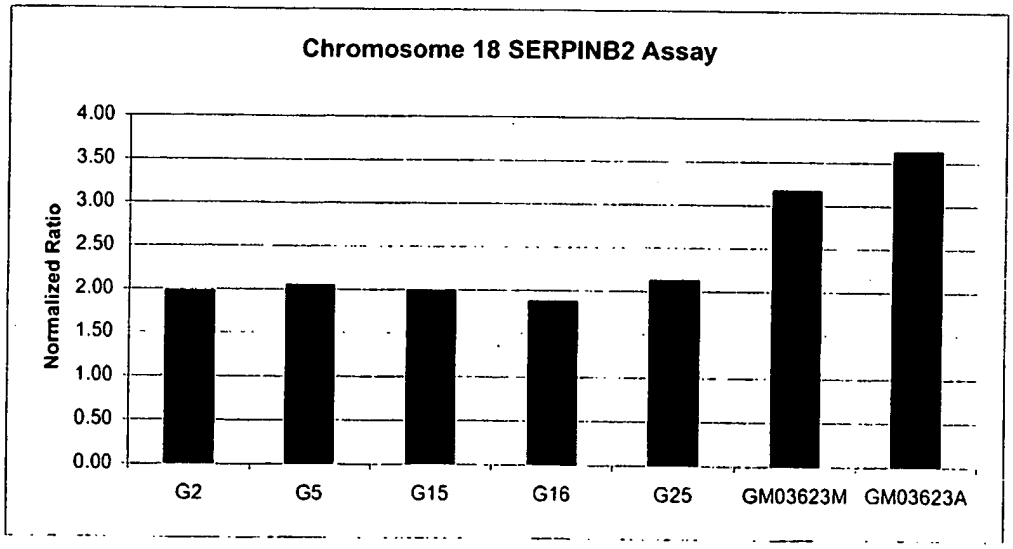

Chromosome 18 SERPINB2 Assay
Normalized Ratio
4.00
3.50
3.00
2.50
2.00
1.50
1.00
0.50
0.00
G2
G5
G15
G16
G25
GM03623M
GM03623A

FIGURE 7

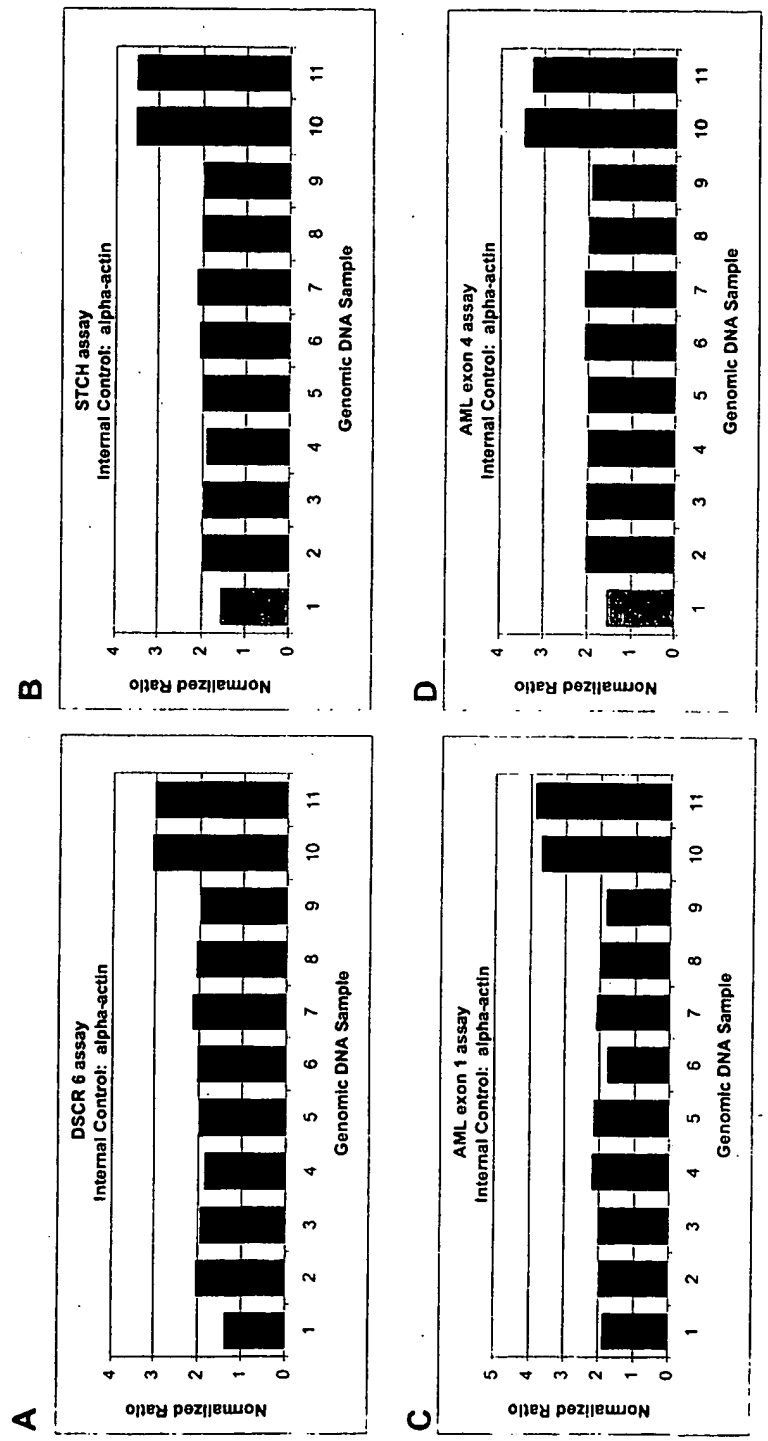
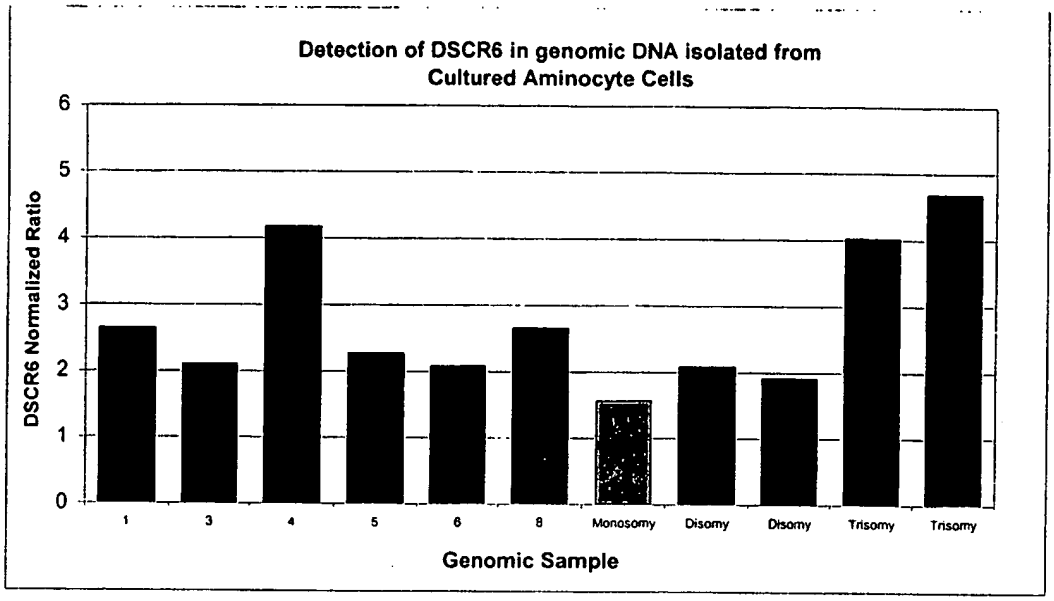

A
DSCR 6 assay
Internal Control: alpha-actin
Normalized Ratio
Genomic DNA Sample
B
STCH assay
Internal Control: alpha-actin
Normalized Ratio
Genomic DNA Sample
C
AML exon 1 assay
Internal Control: alpha-actin
Normalized Ratio
Genomic DNA Sample
D
AML exon 4 assay
Internal Control: alpha-actin
Normalized Ratio
Genomic DNA Sample

FIGURE 7 (continued)

E



F

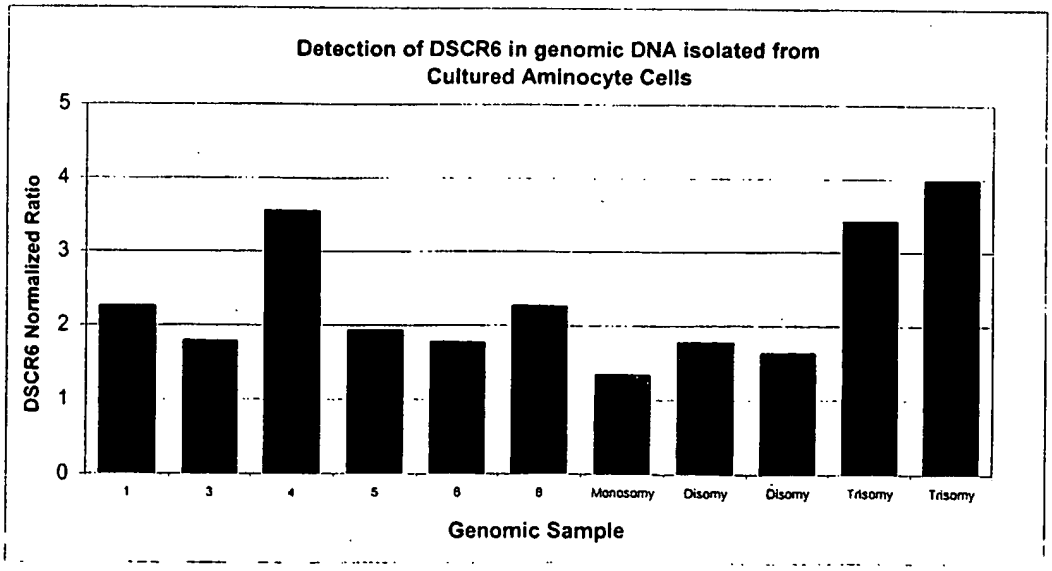
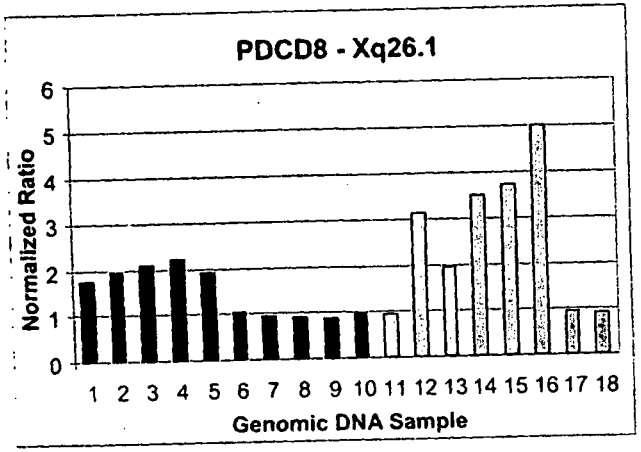
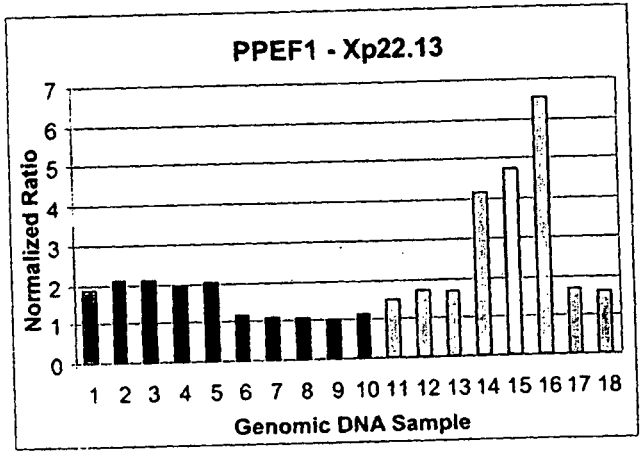


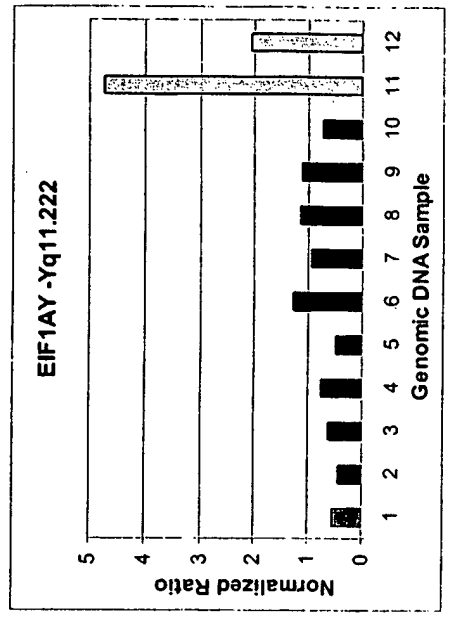
FIGURE 8

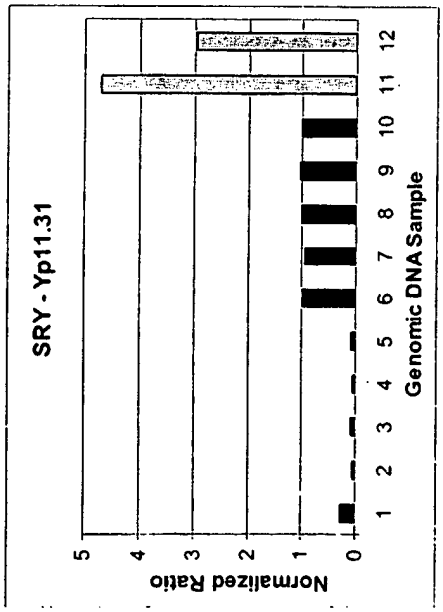




Sample #	Description
1-5	46, XX
6-10	46, XY
11	45,X
12	30% 45, X / 70% 46, X, iX (qter>cen>Qter)
13	40% 45, X / 60% 46, X, iX (qter>cen>Qter)
14	48, XXX, +18
15	48, XXXX
16	48,XXXX / 49,XXXXX
17-18	47, XYY

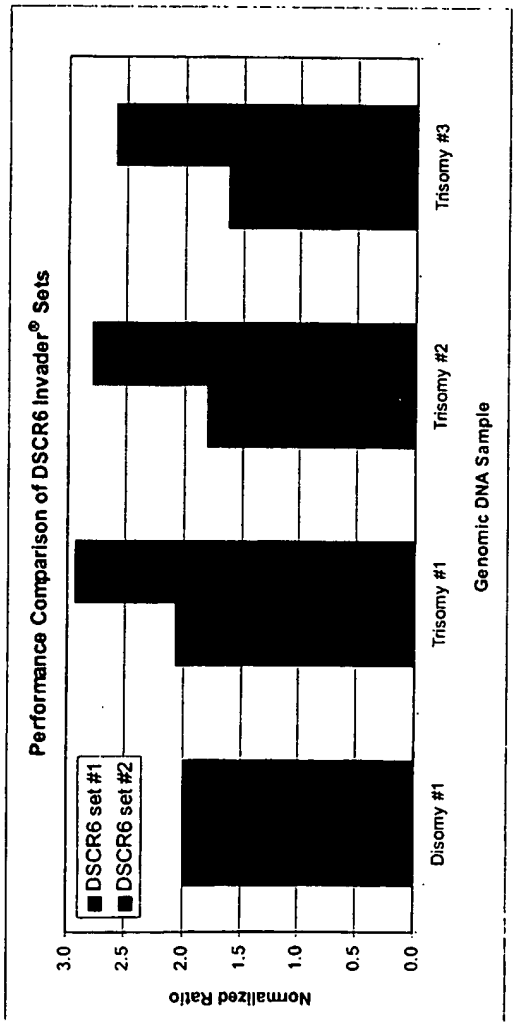
FIGURE 9





Sample #	Description
1-5	46, XX
6-10	46, XY
11-12	47, XYY

FIGURE 10



Trisomy #1: Coriell AG13429

Trisomy #2: Coriell AG09394

Trisomy #3: Coriell AG10098

-all genomic DNA's were prepped using the Gentra Autopure Prep

FIGURE 11A

STCH Signal (FAM)

% Maternal Contamination							0	1	5	10	20	50	80	90	95	99	100	0	1	5	10	20	50	80	90	95	99	100
ng	0	50 #2	50 #3	50 #4	50 #5	0	0	1	5	10	20	50	80	90	95	99	100	0	0.5	2.5	5	10	25	40	45	47.5	49.5	50
ng	0	0	0	0	0	50 #2	100	99	95	90	80	50	20	10	5	1	0	50	49.5	47.5	45	40	25	10	5	2.5	0.5	0
Si	205	450	482	421	457	876	1024	1143	1096	1156	1122	959	766	805	749	750	765	741	795	803	809	781	639	561	553	516	497	534
	235	448	482	418	468	899	1107	1174	1157	1097	1048	940	802	765	758	725	713	803	792	821	777	722	627	578	541	478	497	509
Av	220	449	482	420	463	888	1066	1159	1127	1127	1085	950	784	785	754	738	739	772	794	812	793	752	633	570	547	497	497	522
SD	21	1	0	2	8	16	59	22	43	42	52	13	25	28	6	18	37	44	2	13	23	42	8	12	8	27	0	18
%C	10%	0%	0%	1%	2%	2%	6%	2%	4%	4%	5%	1%	3%	4%	1%	2%	5%	6%	0%	2%	3%	6%	1%	2%	2%	5%	0%	3%
Signal/Bkgd		2.04	2.19	1.91	2.10	4.03	4.84	5.27	5.12	5.12	4.93	4.32	3.56	3.57	3.43	3.35	3.36	3.51	3.61	3.69	3.60	3.42	2.88	2.59	2.49	2.26	2.26	2.37
Net Signal		229	262	200	243	668	846	939	907	907	865	730	564	565	534	518	519	552	574	592	573	532	413	350	327	277	277	302

Alpha Actin Signal (Red)

% Maternal Contamination							0	1	5	10	20	50	80	90	95	99	100	0	1	5	10	20	50	80	90	95	99	100
ng	0	50 #2	50 #3	50 #4	50 #5	0	0	1	5	10	20	50	80	90	95	99	100	0	0.5	2.5	5	10	25	40	45	47.5	49.5	50
ng	0	0	0	0	0	50 #2	100	99	95	90	80	50	20	10	5	1	0	50	49.5	47.5	45	40	25	10	5	2.5	0.5	0
Si	107	309	341	299	322	381	439	436	450	490	528	553	497	562	563	586	594	284	289	323	335	342	342	334	342	345	345	388
	114	303	359	296	330	385	436	438	446	472	479	573	497	504	559	559	560	295	300	319	320	327	328	324	338	304	339	357
Av	111	306	350	298	326	383	438	437	448	481	504	563	497	533	561	573	577	290	295	321	328	335	335	329	340	325	342	373
SD	5	4	13	2	6	3	2	1	3	13	35	14	0	41	3	19	24	8	8	3	11	11	10	7	3	29	4	22
%C	4%	1%	4%	1%	2%	1%	0%	0%	1%	3%	7%	3%	0%	8%	1%	3%	4%	3%	3%	1%	3%	3%	3%	2%	1%	9%	1%	6%
Signal/Bkgd		2.77	3.17	2.69	2.95	3.47	3.96	3.95	4.05	4.35	4.56	5.10	4.50	4.82	5.08	5.18	5.22	2.62	2.67	2.90	2.96	3.03	3.03	2.98	3.08	2.94	3.10	3.37
Net Signal		196	240	187	216	273	327	327	338	371	393	453	387	423	451	462	467	179	184	211	217	224	225	219	230	214	232	262
Ratio		0.59	0.55	0.54	0.57	1.23	1.30	1.44	1.35	1.23	1.11	0.81	0.73	0.67	0.59	0.56	0.58	1.55	1.57	1.41	1.33	1.19	0.92	0.80	0.72	0.65	0.60	0.58
Av	0.56																											
Normalized Ratio		2.10	1.96	1.91	2.02	4.40	4.64	5.16	4.82	4.39	3.95	2.89	2.62	2.40	2.13	2.01	2.00	5.53	5.59	5.05	4.74	4.26	3.30	2.87	2.56	2.32	2.15	2.07

FIGURE 11B

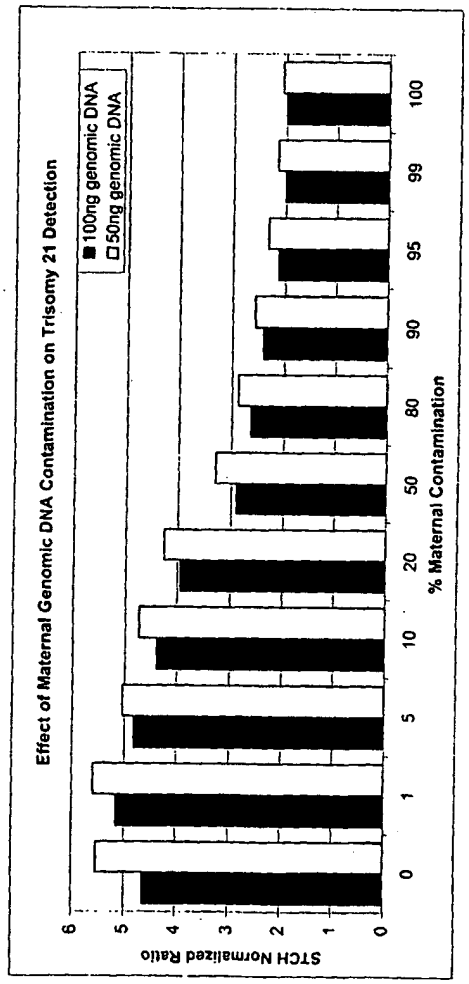
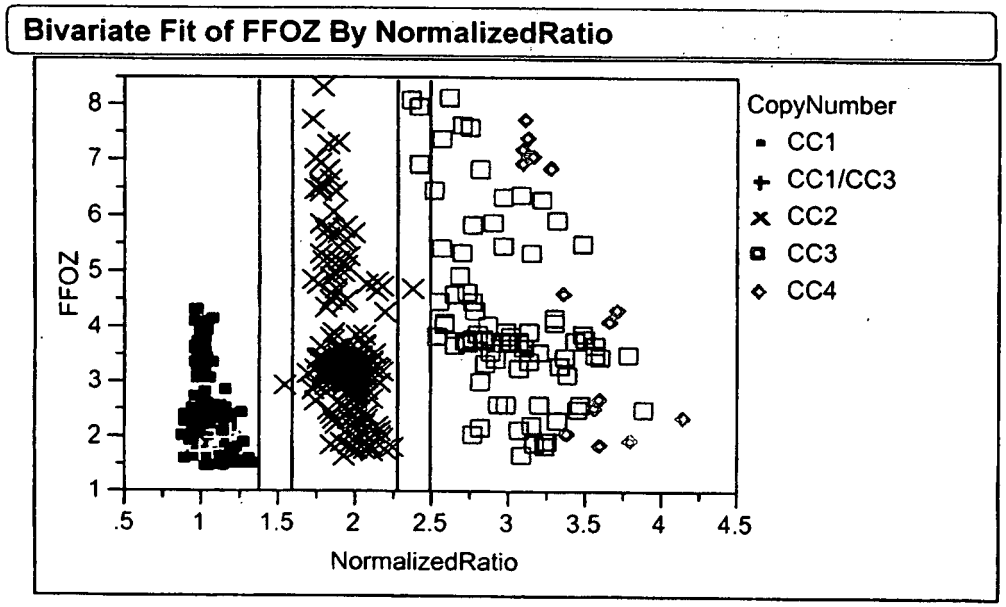

Effect of Maternal Genomic DNA Contamination on Trisomy 21 Detection
100ng genomic DNA
50ng genomic DNA
STCH Normalized Ratio
0
1
2
3
4
5
6
% Maternal Contamination
0
1
5
10
20
50
80
90
95
99
100

Figure 12

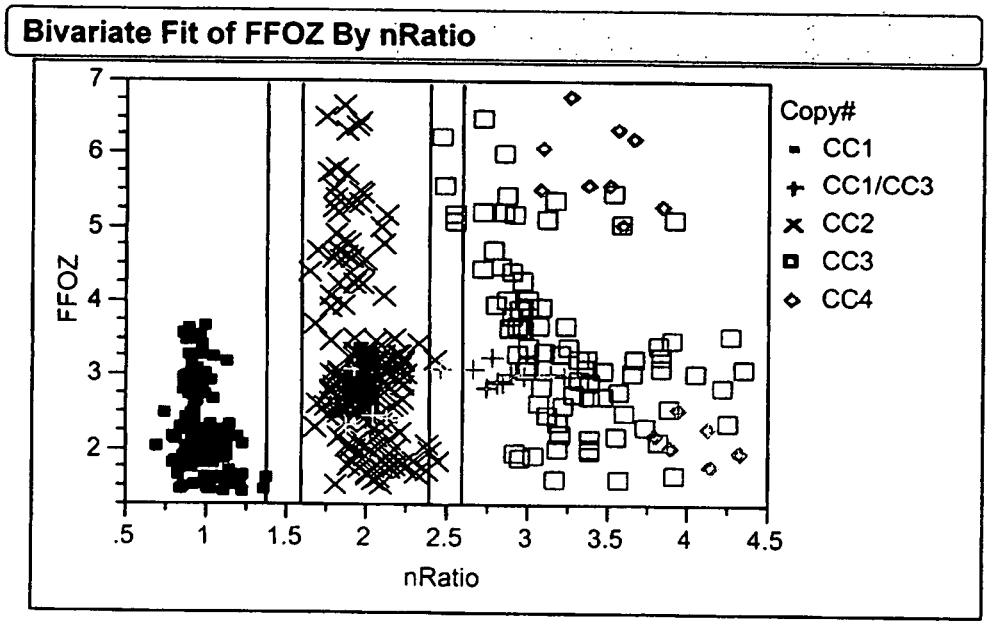
Chrom	Gene	Accession #	Cytoband	Target Sequence	SEQ ID NO:
1	alpha actin (ACTA1)	M20543	1q42.13	(ATGTCACAGGGTTCC[T]AACAGGCCACTCTTCCCTGGATGG)	203
1	HIST2H2BE	NM_003528	1q21.2	GTATCCACAGGA(GGCCCCATCGCACGGCAGAACT[G]GTGACCGGACACA)AAAGCTGCTGCCAAGCGTCAGTCATACAGAGCTGTCACGCAGT	204
13	DLEU1	NM_005887	13q14.2	G(GCACATGCGCAGA[A]TCATCGTGGTGCACGGCTCTCCCT)TTGCTTCTTCGGTTGCAGTCCTCTTGCTTCTTGCGCGTGCGTGTAGCGCTTT	205
13	PCDH9	NM_020403	13q21.33	ATGGTAACCGTTGC(TCGAGCAGGAGTG[G]AGCTGCCGTCACTAGCCAGCACTGT)CACTTTGTGAATGGCTGTCTCCTCTCTATCTAAGGACC	206
18	FLJ23403	NM_022068	18p11.22	GCCAACACTTCAGGATGAATATGTGAGCATAGATGTCCT(CCACACAGATCCAG[C]TGGACAGGCTCAAAGTTGTGTCCGTCCA)CACCCAGTC	207
X	PFKFB1	NM_002625	Xp11.21	ATGGTCCTCTTCATGTGACTGGTCCA[CACCTTCAGGGAGCT[G]ATGCCCTGGGACTGAATGAAGTTGGCCA]GGGCATAGGCA	208
Y	PRKY	NM_002760	Yp11.2	CTCCTTCT(GCACCCAGAGTAGGGGTGGAGTATACCCTT[G]CCATGTGTTAGTGAAAATG)TGTGCAGTGAGAAGCCAGTTGGGTCCCCTCGGC	209
21	NRIP1	NM_003489	21q11.2	GGCTCCGATTTAAAGTCTTCG(GACACTGGTAAGGCA[G]TGCGCTTCTCTGCACAGCAGGAGC)CATACCCAAGAATGGGGCACTCTTAGCAT	210
21	HLCS	NM_000411	21q22.13	GAGGATG(AGGTTTCTCAGCATGT[C]GAAGGAGTTGCCGTCCGGGTGCA)CAGTCACAACCTCGCCGCCCTCCTGGTGAACCTGGAGGAAGCCA	211
18	CN2	NM_018235	18q22.3	TGGCC(GGAGAAGAGAGGCGAAATCAGGAGGATGATGGAA[G]TTGCTGCTGCAGAT)GTTAAGCAGTTGGGGGGCTCTGTGGAACTGGTGGATA	212
X	MTMR8	NM_017677	Xq11.2	CCTTGGGTACCGTAATATGAT(CCATGACTGCAGTTCCCGCCAC[C]GGAAGATCTCAGTGCT)ACTCCAGATGCCGCCGCCACCGGTCTAGCCG	213
X	FLJ21174	NM_024863	Xq22.2	GAAGAGTTCAGCTGCTAGGTTCTTATTTTG(GGACTCTCATGATGCT[G]AGGTCAAGATGCCAGCTGGAGGCCA)GGCGCCGCAGCTCACGCCT	214
X	PCTK1	NM_033018	Xp11.3	GAGATTGTGCACGAGGACTTGAAGATGGGGTCTGATGGGGAG(AGTGACCAGGCTTC[A]GCCACGTCCTCGGATGAGGTGCAG)TCTCCAGTGA	215
Y	SRY	NM_003140	Yp11.31	TCATCCCTGTACAACCTGTTGTCCAGTTGCACTTCGCT(GCAGAGTACCGAAG[C]GGGATCTGCGGGAAGCAAACTGCAATTCTTC)GGCAGCA	216

Figure 13A
Chromosome Xp Invader Assay:PFKFB1+PCTK1 –Varying DNA Levels



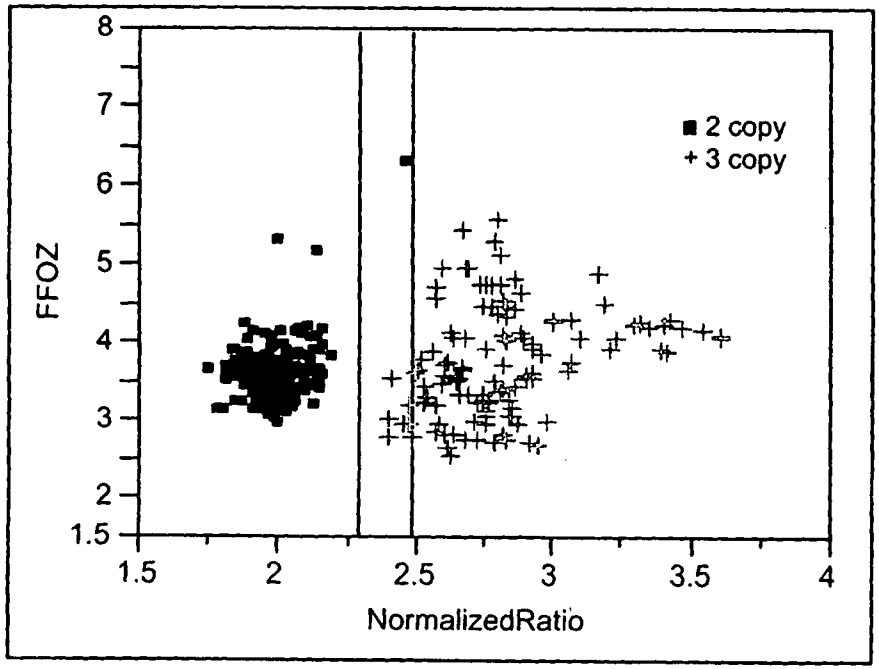
- 3 - 30 ng of DNA per reaction
- Equivocol zones: 1.4 -1.6; 2.3 – 2.5
- N=637 samples, 517 normal, 120 aneuploid
- No call rate 1.9% (7 samples < 1.4 FOZ, 5 equivocal samples)
- Miscall rate 0%

Figure 13B
Chromosome Xq Invader Assay:MTMR8+FLJ21174 –Varying DNA Levels



- 3 - 30 ng of DNA per reaction
- Equivocol zones: 1.4 -1.6; 2.4 – 2.6
- N=638 samples, 518 normal, 120 aneuploid
- No call rate 3.3% (15 samples < 1.4 FOZ, 6 equivocal samples)
- Miscall rate 0%

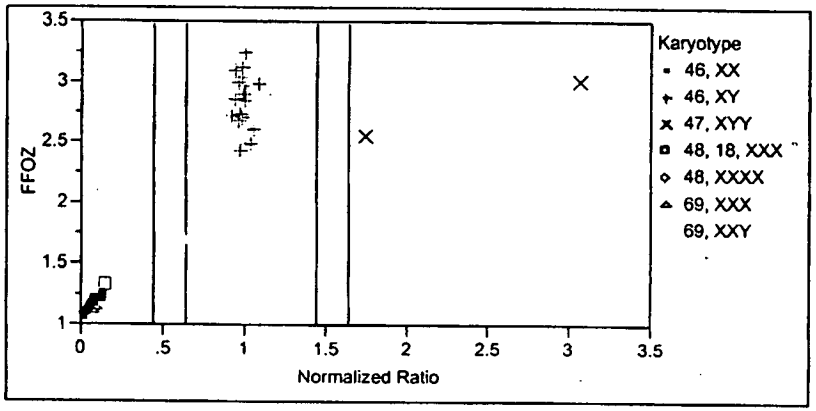
Figure 14. Chromosome 18 Invader® Assay – Sample Mixtures



- Trisomy 18 / Disomy sample mixtures
- 10 ng of DNA per reaction
- Equivocal zone: 2.3 – 2.5
- N=315 samples, 198 disomy, 117 trisomy (+18 with 0%, 10% and 20% disomy contamination)
- No call rate 2.9%
- Miscall rate 0%

Figure 15: Analysis of Triploidy Samples (69, XXY)

Normalized Ratio Analysis

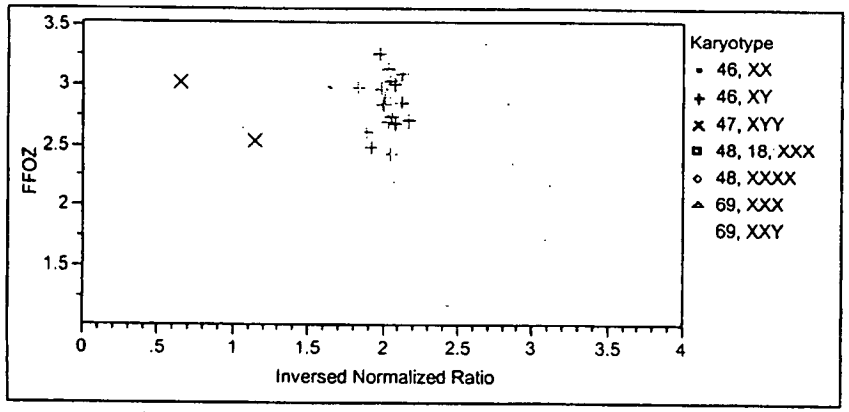


Ratio = ((Net chromosome specific FOZ))
_____(Net internal control FOZ)

where Net FOZ = FOZ – 1

Normalized Ratio= (Ratio of unknown sample) / (Ratio of male control sample) X (1)

Inversed Normalized Ratio Analysis



Inversed Ratio = ((Net internal control FOZ))
_____(Net chromosome specific FOZ)

where Net FOZ = FOZ – 1

Inversed Normalized Ratio=

(Ratio of unknown sample) / (Ratio of male control sample) X (2)

Note: Samples not containing a Y chr. not shown on graph due to high inversed normalized ratios (>10)